Human Genetics

Volume 64 1983

Editorial Board

W. Lenz, Münster/W.
A. G. Motulsky, Seattle
F. Vogel, Heidelberg
U. Wolf, Freiburg i. Br.

Advisory Board

G. Anders, Groningen
H. Bickel, Heidelberg
N. P. Bochkov, Moscow
D. Bootsma, Rotterdam
K. H. Degenhardt, Frankfurt/Main
B. Dutrillaux, Paris
C. J. Epstein, San Francisco
G. Flatz, Hannover
M. Fraccaro, Pavia
U. Francke, New Haven
W. Fuhrmann, Giessen
H. Hoehn, Würzburg
P. A. Jacobs, Honolulu
W. Jaeger, Heidelberg
Y. W. Kan, San Francisco
W. Krone, Ulm
V. A. McKusick, Baltimore
E. Matsunaga, Misima
M. Mikkelsen, Glostrup
O. J. Miller, NewYork
E. Passarge, Essen
H. Ritter, Tübingen
D. F. Roberts, Newcastle/Tyne
W. Schmid, Zürich
U. W. Schnyder, Zürich
H. G. Schwarzacher, Wien
C. R. Scriver, Montreal
K. Sperling, Berlin
R. Williamson, London

Springer International

Human Genetics

Human Genetics was founded in 1964 and published up to Vol. 30 as Humangenetik–Human Genetics–Génétique humaine.

Copyright

Submission of a manuscript implies that the work described has not been published before (except in the form of an abstract or as part of a published lecture, review or thesis), that it is not under consideration for publication elsewhere, that its publication has been approved by all the authors and by the responsible authorities–tacitly or explicitly–in the laboratories where the work was carried out and that, if accepted, it will not be published elsewhere in the same form, in either the same or another language, without the consent of the copyright holders. By submitting a manuscript, the authors agree that the copyright for their article is transferred to the publisher if and when the article is accepted for publication. The copyright covers the exclusive rights to reproduce and distribute the article, including reprints, photographic reproductions, microform, electronic data base, video disk, or any other reproductions of similar nature and translations.

Photographic reproduction, microform, electronic data base, video disk, or any other reproduction of text, figures, or tables from this journal is prohibited without permission obtained from the publisher.

The use of general descriptive names, trade names, trade marks, etc., in this publication, even if the former are not specifically identified, is not to be interpreted as exempt from the relevant protective laws and regulations.

While the advice and information in this journal is believed to be true and accurate at the date of its going to press, neither the authors nor the editors nor the publisher can accept any legal responsibility for errors or omissions that may have been made. The publisher makes no warranty, express or implied, with respect to the material contained herein.

Special Regulations for the USA

The Article Fee Code on the first page of an article in this journal indicates the copyright owner's consent that in the USA copies may be made for personal or internal use, provided the stated fee for copying beyond that permitted by Sections 107 or 108 of the United States Copyright Law is paid through the Copyright Clearance Center, Inc., 21 Congress Street, Salem, MA 01970, U.S.A. If a code does not appear, copies of the article may be made without charge, provided permission is obtained from the publisher. The copyright owner's consent does not extend to copying for general distribution, for promotion, for creating new works, or for resale. Specific written permission must be obtained from the publisher for such copying.

Other Regulations

Authors of this journal can benefit from library and photocopy fees collected by VG WORT if certain conditions are met. If an author lives in the Federal Republic of Germany or in West Berlin it is recommended that he contacts Verwertungsgesellschaft WORT, Abteilung Wissenschaft, Goethestraße 49, D-8000 München 2, for detailed information.

Printed in Germany
by Petersche Druckerei GmbH & Co. Offset KG,
Rothenburg ob der Tauber

© Springer-Verlag Berlin · Heidelberg 1983

Contents

Abbas, H., Ahmad, M.: Persistence of high intestinal lactase activity in Pakistan (Orig. invest.) . . . 277
Abel, G., Schimmer, O.: Induction of structural chromosome aberrations and sister chromatid exchanges in human lymphocytes in vitro by aristolochic acid (Orig. invest.) . 131
Abrisqueta, J. A., s. Martín, M. J. . . . 100
Ahmad, M., s. Abbas, H. . . . 277
Ahuja, Y. R., s. Jaju, M., et al. . . . 42
Airhart, S. D., s. Nussbaum, R. L., et al. . . . 148
Albert, D. M., s. Dryja, T. P., et al. . . . 151
Andersen, O., s. Tabor, A., et al. . . . 196
Anderson, C. E., s. Sparkes, R. S., et al. . . . 146
Aquirre, A. I., s. Martínez de Pancorobo, M., et al. . . . 395
Atkin, N. B.: Endomitosis in human trophoblast? (Letter to the editor) . . . 309
Auconi, P., s. Pascali, V. L. . . . 232
Aurias, A., s. Iselius, L., et al. . . . 343

Baeteman, M. A., s. Balestrazzi, P., et al. . . . 305
Balakirev, S. A., s. Fleischman, E. W., et al. . . . 254
Balestrazzi, P., Baeteman, M. A., Mattei, M. G., Mattei, J. F.: Franceschetti syndrome in a child with a de novo balanced translocation (5;13)(q11;p11) and significant decrease of hexosaminidase B (Cases observed) . . . 305
Barbi, G., s. Steinbach, P., et al. . . . 279
Barneveld, R. A., Keijzer, W., Tegelaers, F. P. W., Ginns, E. I., Geurts van Kessel, A., Brady, R. O., Barranger, J. A., Tager, J. M., Galjaard, H., Westerveld, A., Reuser, A. J. J.: Assignment of the gene coding for human β-glucocerebrosidase to the region q21–q31 of chromosome 1 using monoclonal antibodies (Orig. invest.) . . . 227
Barranger, J. A., s. Barneveld, R. A., et al. . . . 227
Bartelheimer, H., s. Rüdiger, H. W., et al. . . . 407
Bastard, C., s. Iselius, L., et al. . . . 343
Bauchinger, M., s. Rauscher, K. H. . . . 73
Baur, S., s. Steinbach, P., et al. . . . 279
Beauregard, L., s. Tantravahi, U., et al. . . . 33
Beck, B., s. Brøndum Nielsen, K., et al. . . . 240
Bender, K., s. Mauff, G., et al. . . . 186
Bender, K., s. Wieacker, P., et al. . . . 143
Berghe, H. Van den, s. Fryns, J. P., et al. . . . 194
Bhagat, C. I., s. Dempsey, J. L., et al. . . . 288
Biagioli, P., s. Franceschini, P., et al. . . . 97,98
Bianco, R., s. Franceschini, P., et al. . . . 97,98
Blom, W., s. Gemund, J. J. van, et al. . . . 50
Blumhardt, R., s. Quattromani, F., et al. . . . 116
Bonifaci, E., s. Fraccaro, M., et al. . . . 388
Bostock, C., s. Mayer, H., et al. . . . 283
Bottelli, A. M., s. Iselius, L., et al. . . . 343
Brackertz, M., Kubbies M., Feige, A., Salk, D.: Decreased oxygen supply enhances growth in culture of human midtrimester amniotic fluid cells (Orig. invest.) . . . 334
Brady, R. O., s. Barneveld, R. A., et al. . . . 227
Breyel, E., s. Mayer, H., et al. . . . 283
Brøndum Nielsen, K., Tommerup, N., Poulsen, H., Jacobsen, P., Beck, B., Mikkelsen M.: Carrier detection and X-inactivation studies in the fragile X syndrome. Cytogenetic studies in 63 obligate and potential carriers of the fragile X (Orig. invest.) . . . 240
Bruns, G. A. P., s. Dryja, T. P., et al. . . . 151
Bühler, E., s. Fraccaro, M., et al. . . . 388
Bühler, E. M., Bühler, U. K., Christen, R.: Terminal or interstitial deletion in chromosome 8 long arm in Langer-Giedion syndrome (TRP II syndrome)? (Orig. invest.) . . 163
Bühler, U. K., s. Bühler, E. M., et al. . . . 163
Bui, T.-H., s. Iselius, L., et al. . . . 343
Burck, U.: Genetic aspects of hemifacial microsomia (Clinical case report) . . . 291
Burk, R. D., Stamberg, J., Young, K. E., Smith, K. D.: Use of repetitive DNA for diagnosis of chromosomal rearrangements (Orig. invest.) . . . 339
Buytaert, C., s. Elsen, A. F. Van, et al. . . . 235

Callen, P. W., s. Stephens, J. D., et al. . . . 191
Carrel, R. E., s. Wulfsberg, E. A., et al. . . . 271
Caufin, D., s. Fraccaro, M., et al. . . . 388
Caufin, D., s. Iselius, L., et al. . . . 343
Christen, R., s. Bühler, E. M., et al. . . . 163
Cignacco, G., s. Fraccaro, M., et al. . . . 388
Cirillo Silengo, M., s. Franceschini, P., et al. . . . 97,98
Cleve, H., s. Görg, A., et al. . . . 222
Cochet, C., s. Turleau, C., et al. . . . 65
Cohen, G., Manuel A., Cohen, M., Endrey-Walder, S., McBride, W.: Terminal rearrangement of chromosomes 21 detected in amniotic fluid, resulting in a trisomy 21 (Cases observed) . . . 203
Cohen, G., Manuel, A., Cohen, M., Fagan, K., Grunstein, H.: A deletion of heterochromatin only of the Y chromosome in an azoospermic male (Clinical case report) . . . 297
Cohen, M., s. Cohen, G., et al. . . . 203, 297
Cong, N. Van, s. Grandchamp, B., et al. . . . 180
Cooper, D. N.: Eukaryotic DNA methylation (Review article) 315
Cox, D. R., Palmiter, R. D.: The metallothionein-I Gene maps to mouse chromosome 8: Implications for human Menkes' disease (Orig. invest.) . . . 61
Créau-Goldberg, N., s. Turleau, C., et al. . . . 65
Cremer, C., s. Müller, C. R., et al. . . . 110
Crist, M., s. Sparkes, R. S., et al. . . . 146
Cross, P. K., s. Hook, E. B. . . . 267
Czeizel, A.: New lethal omphalocele-cleft palate syndrome? (Cases observed) . . . 99
Czeizel, A., Flatz, G., Flatz, S. D.: Prevalence of primary adult lactose malabsorption in Hungary (Orig. invest.) . . . 398

Dallapiccola, B., s. Wieacker, P., et al. . . . 143
Dalprà, L., s. Iselius, L., et al. . . . 343
Das, B. C., Sharma, T.: Reduced frequency of baseline sister chromatid exchanges in lymphocytes grown in antibiotics and serum-excluded culture medium (Orig. invest.) . . . 249
Davi, G., s. Franceschini, P., et al. . . . 97,98
David, V., Fauchet, R., Phengsavath, H., Guenet, L., Le Gall, J. Y.: Properdin factor B (Bf) polymorphism: Subtyping of SS phenotypes (Short comm.) . . . 189
Davies, K. E., s. Müller, C. R., et al. . . . 110
Davies, K. E., s. Wieacker, P., et al. . . . 143
Delendi, N., s. Fraccaro, M., et al. . . . 388
Delendi, N., s. Iselius, L., et al. . . . 343
Dempsey, J. L., Morley, A. A., Seshadri, R. S., Emmerson, B. T., Gordon, R., Bhagat, C. I.: Detection of the carrier state for an X-linked disorder, the Lesch-Nyhan syndrome, by the use of lymphocyte cloning (Short comm.) . . . 288
Dipierri, J. E., Fraisse, J.: A simple combined Ag-I/RHG technique for human metaphase chromosomes (Short comm.) 286
Djalali, M., s. Steinbach, P. . . . 402
Dopfer, R., s. Teufel, M., et al. . . . 94
Dreyer, M., s. Rüdiger, H. W., et al. . . . 407
Drosdova, T. S., s. Puchkova, G. P., et al. . . . 257
Dryja, T. P., Bruns, G. A. P., Gallie, B., Petersen, R., Green, W., Rapaport, J. M., Albert, D. M., Gerald, P. S.: Low incidence of deletion of the esterase D locus in retinoblastoma patients (Orig. invest.) . . . 151
Dutrillaux, B., s. Iselius, L., et al. . . . 343

Eerdmans, R. F., s. Gemund, J. J. van, et al. . . . 50
Elsen, A. F. Van, Leroy, J. G., Wauters, J. G., Willems, P. J., Buytaert, C., Verheyen, K.: In vitro expression of α-L-fucosidase activity polymorphism observed in plasma (Orig. invest.) . . . 235
Emmerson, B. T., s. Dempsey, J. L., et al. . . . 288
Enders, H., s. Teufel, M., et al. . . . 94
Endrey-Walder, S., s. Cohen, G., et al. . . . 203
Engel, W., s. Schmid, M., et al. . . . 363
Eriksson, A. W., s. Kwee, M. L., et al. . . . 384

Fagan, K., s. Cohen, G., et al. . . . 297
Fauchet, R., s. David, V., et al. . . . 189

Feige, A., s. Brackertz, M., et al. 334
Filly, R. A., s. Stephens, J. D., et al. 191
Fitzgerald, P. H., Stewart, J., Suckling, R. D.: Retinoblastoma mutation rate in New Zealand and support for the two-hit model (Orig. invest.) 128
Flatz, G., s. Czeizel, A., et al. 398
Flatz, S. D., s. Czeizel, A., et al. 398
Fleischer-Reischmann, B., s. Wunder, E. 167
Fleischman, E. W., Prigogina, E. L., Iljinskaja, G. W., Konstantinova, L. N., Puchkova, G. P., Volkova, M. A., Frenkel, M. A., Balakirev, S. A.: Chromosomal rearrangements with a common breakpoint at 6p23 in five cases of myeloid leukemia (Orig. invest.) 254
Fleischman, E. W., s. Puchkova, G. P., et al. 257
Fonatsch, C., Schwinger, E.: Frequency of fragile X chromosomes, fra(X), in lymphocytes in relation to blood storage time and culture techniques (Orig. invest.) 39
Foubert, Ch., s. Grandchamp, B., et al. 180
Fraccaro, M., Iselius, L., et al. 343
Fraccaro, M., Zuffardi, O., Bühler, E., Schinzel, A., Simoni, G., Witkowski, R., Bonifaci, E., Caufin, D., Cignacco, G., Delendi, N., Gargantini, L., Losanowa, T., Marca, L., Ullrich, E., Vigi, V.: Deficiency, transposition, and duplication of one 15q region may be alternatively associated with Prader-Willi (or a similar) syndrome. Analysis of seven cases after varying ascertainment (Orig. invest.) . . 388
Fraisse, J., s. Dipierri, J. E., ... 286
Franceschini, P., Cirillo Silengo, M., Davi, G., Bianco, R., Biagioli, M.: Interstitial deletion of the long arm of chromosome 3 in a patient with mental retardation and congenital anomalies (Cases observed) 97
Franceschini, P., Cirillo Silengo, M., Davi, G., Bianco, R., Biagioli, P.: Interstitial deletion of the long arm of chromosome 2 (q31q33) in a girl with multiple anomalies and mental retardation (Cases observed) 98
Frenkel, M. A., s. Fleischmann, E. W., et al. 254
Frézal, J., Munnich, A., Mitchell, G.: One gene, several messages. From multifunctional proteins to endogenous opiates (Review article) 311
Friedl, W., s. Propping, P. 105
Froesch, E. R., s. Schoenle, E., et al. 86
Froesch, P., s. Schoenle, E., et al. 86
Fryns, J. P., Heremans, G., Marien, J., Berghe, H. Van den: Langer-Giedion syndrome and deletion of the long arm of chromosome 8. Confirmation of the critical segment to 8q23 (Clinical case report) 194
Fryns, J. P., s. Petit, P. 303
Fukushima, Y., s. Iselius, L., et al. 343
Fukushima, Y., Kuroki, Y., Izawa T.: Two cases of the Langer-Giedion syndrome with the same interstitial deletion of the long arm of chromosome 8: 46,XY or XX,del(8)(q23.3q24.13) (Clinical case report) 90

Galjaard, H., s. Barneveld, R. A., et al. 227
Gall, J. Y. Le, s. David, V., et al. 189
Gallie, B., s. Dryja, T. P., et al. 151
Gargantini, L., s. Fraccaro, M., et al. 388
Gedde-Dahl, T., Jr., s. Schamaun, O., et al. 380
Gemund, J. J. van, Giesberts, M. A. H., Eerdmans, R. F., Blom, W., Kleijer, W. J.: Morquio-B disease, spondyloepiphyseal dysplasia associated with acid β-galactosidase deficiency. Report of three cases in one family (Orig. invest.) 50
Geraedts, J. P. M., s. Iselius, L., et al. 343
Gerald, P. S., s. Dryja, T. P., et al. 151
Geurts van Kessel, A., s. Barneveld, R. A., et al. 227
Giesberts, M. A. H., s. Gemund, J. J. van, et al. 50
Ginns, E. I., s. Barneveld, R. A., et al. 227
Giraud, F., s. Mattei, J. F., et al. 356
Görg, A., Weser, J., Westermeier, R., Postel, W., Weidinger, S., Patutschnick, W., Cleve, H.: Isoelectric focusing with immobilized pH gradients for the analysis of transferrin (Tf) subtypes and variants (Orig. invest.) 222
Golbus, M. S., s. Stephens, J. D., et al. 191
Goldman, E., s. Winter, R. M., et al. 156
Goos, C. M. A. A., s. Miguel, A., et al. 176
Gordon, R., s. Dempsey, J. L., et al. 288
Gottschall, A., Lošan, F., Pelz, L., Wiedersberg, H.: A new type of familial chromosome translocation involving 3p and 6q in two unrelated families (Cases observed) . . . 304
Grandchamp, B., Weil, D., Nordmann, Y., Cong, N. Van, Verneuil, H. de, Foubert, Ch., Gross, M.-S.: Assignment of the human coproporphyrinogen oxidase to chromosome 9 (Short comm.) 180
Gray, J. W., s. Müller, C. R., et al. 110
Green, W., s. Dryja, T. P., et al. 151
Gross, M.-S., s. Grandchamp, B., et al. 180
Grouchy, J. De, s. Iselius, L., et al. 343
Grouchy, J. de, s. Turleau, C., et al. 65
Grundstein, H., s. Cohen, G., et al. 297
Guenet, L., David, V., et al. 189
Güttler, F., s. Tønnesen, T., et al. 371
Gyftodimou, J., s. Iselius, L., et al. 343

Halbrecht, I., s. Shabtai, F., et al. 273
Hanley, A. L., s. Iselius, L., et al. 343
Hansmann, I., s. Iselius, L., et al. 343
Hart, J., s. Shabtai, F., et al. 273
Hatano, S., s. Yoshimitsu, K., et al. 200
Hayashidani, M., s. Yoshimitsu, K., et al. 200
Heremans, G., s. Fryns, J. P., et al. 194
Herrera, C., s. Kang, E. S., et al. 80
Hitzig, W. H., Seger, R. A.: Chronic granulomatous disease, a heterogeneous syndrome (Review article) 207
Hörler, H., s. Vogel, W., et al. 246
Hook, E. B., Cross, P. K.: Spontaneous abortion and subsequent Down syndrome livebirth (Orig. invest.) 267
Hook, E. B., Warburton, D.: The distribution of chromosomal genotypes associated with Turner's syndrome: Livebirth prevalence rates and evidence for diminished fetal mortality and severity in genotypes associated with structural X abnormalities or mosaicism (Orig. invest.) 24
Horn, N., s. Wieacker, P., et al. 139
Horst, J., s. Oehme, R., et al. 376
Horst, J., Oehme, R., Kleihauer, E., Kohne, E.: DNA restriction mapping identifies the chromosome carrying the mutant Hb Presbyterian β-globin gene (Orig. invest.) . . 263

Iljinskaja, G. W., s. Fleischman, E. W., et al. 254
Inoue, A., s. Motegi, T., et al. 160
Iselius, L., Lindsten, J., Aurias, A., Fraccaro, M., Bastard, C., Bottelli, A. M., Bui, T.-H., Caufin, D., Dalprà, L., Delendi, N., Dutrillaux, B., Fukushima, Y., Geraedts, J. P. M., Grouchy, J. De, Gyftodimou, J., Hanley, A. L., Hansmann, I., Ishii, T., Jalbert, P., Jingeleski, S., Kajii, T., Koskull, H. von, Niikawa, N., Noel, B., Pasquali, F., Probeck, H. D., Robinson, A., Roncarati, E., Sachs, E., Scappaticci, S., Schwinger, E., Simoni, G., Veenema, H., Vigi, V., Volpato, S., Wegner, R.-D., Welch, J. P., Winsor, E. J. T., Zhang, S., Zuffardi, O.: The 11q;22q translocation: A collaborative study of 20 new cases and analysis of 110 families (Orig. invest.) 343
Ishii, T., s. Iselius, L., et al. 343
Isusquiza, M., s. Martínez de Pancorbo, M., et al. 395
Iwasaki, M., s. Nakasono, I., et al. 184
Izawa, T., s. Fukushima, Y., et al. 90

Jacobsen, P., s. Brøndum Nielsen, K., et al. 240
Jaju, M., s. Jaju, M., et al. 42
Jaju, M., Jaju, M., Ahuja, Y. R.: Cytogenetic effects of chemotherapy with three combinations of anti-tubercular drugs involving isoniazid, thiacetazone, para-aminosalicylic acid and streptomycin on human lymphocytes: Chromosome aberrations. Sister chromatid exchanges and mitotic index (Orig. invest.) 42
Jalbert, P., s. Iselius, L., et al. 343
Jingeleski, S., s. Iselius, L., et al. 343
Joenje, H., s. Kwee, M. L., et al. 384
Jorgenson, R. J., s. Quattromani, F., et al. 116

Juberg, R. C.: Origin of chromosomal abnormalities: Evidence for delayed fertilization in meiotic nondisjunction (Orig. invest.) . . . 122
Juberg, R. C., Stallard, R., Mowrey, P., Valido, C. L.: Dissociation of a t(12;21) resulting in a normal cell line in two trisomic 21 sons of a nonmosaic t(12;21) father? (Orig. invest.) . . . 216

Kadowaki, H., s. Motegi, T., et al. . . . 160
Kaga, M., s. Motegi, T., et al. . . . 160
Kajii, T., s. Iselius, L., et al. . . . 343
Kamboh, M. I., Kirk, R. L.: Investigation of $PGM_1 3$, $PGM_1 6$, and $PGM_1 7$ variants by isoelectric focussing. Evidence for new subtypes of the PGM_1^3 and PGM_1^7 Alleles (Orig. invest.) . . . 58
Kamp, J. J. P. van de, s. Kwee, M. L., et al. . . . 384
Kang, E. S., Seyer, J., Todd, T. A., Herrera, C.: Variability in the phenotypic expression of abnormal sarcosine metabolism in a family (Orig. invest.) . . . 80
Kantner, G., s. Wettke-Schäfer, R. . . . 1
Kaplan, J. C., s. Reghis, A., et al. . . . 173
Keats, B.: Genetic mapping: X chromosome (Orig. invest.) . 28
Keijzer, W., s. Barneveld, R. A., et al. . . . 227
Kirk, R. L., s. Kamboh, M. I. . . . 58
Kirschner, D. A., s. Tantravahi, U., et al. . . . 33
Klar, D., s. Shabtai, F., et al. . . . 273
Kleihauer, E., s. Horst, J., et al. . . . 263
Kleihauer, E., s. Oehme, R., et al. . . . 376
Kleijer, W. J., s. Gemund, J. J. van, et al. . . . 50
Klisak, I. J., s. Wulfsberg, E. A., et al. . . . 271
Knapp, A., s. Pteruschka, L., et al. . . . 404
Kobayashi, Y., s. Yoshimitsu, K., et al. . . . 200
Koch, M., s. Zakrzewski, S., et al. . . . 55
Kohne, E., s. Horst, J., et al. . . . 263
Kohne, E., s. Oehme, R., et al. . . . 376
Komatsu, M., s. Motegi, T., et al. . . . 160, 205
Koning, H. de, s. Kwee, M. L., et al. . . . 384
Konstantinova, L. N., s. Fleischman, E. W., et al. . . . 254
Koskull, H. von, s. Iselius, L., et al. . . . 343
Krishnamoorthy, R., s. Reghis, A., et al. . . . 173
Kubbies, M., s. Brackertz, M., et al. . . . 334
Kühnau, J., s. Rüdiger, H. W., et al. . . . 407
Kunkel, L., s. Tantravahi, U., et al. . . . 33
Kuroki, Y., s. Fukushima, Y., et al. . . . 90
Kwee, M. L., Poll, E. H. A., Kamp, J. J. P. van de, Koning, H. de, Eriksson, A. W., Joenje, H.: Unusual response to bifunctional alkylating agents in a case of Fanconi anaemia (Orig. invest.) . . . 384

Lambert, W. C. (Cases observed) . . . 204
Latt, S., s. Tantravahi, U., et al. . . . 33
Ledbetter, D. H., s. Nussbaum, R. L., et al. . . . 148
Leroy, J. G., s. Elsen, A. F. Van, et al. . . . 235
Lindsten, J., s. Iselius, L., et al. . . . 343
Lösan, F., s. Gottschall, A., et al. . . . 304
Losanowa, T., s. Fraccaro, M., et al. . . . 388
Lostanlen, D., s. Reghis, A., et al. . . . 173
Lostao, C. M., s. Martínez de Pancorbo, M., et al. . . . 395
Lundsteen, C., s. Tabor, A., et al. . . . 196
Lykkelund, C., s. Tønnesen, T., et al. . . . 371

Machill, G., s. Pteruschka, L., et al. . . . 404
Mahler, M., s. Schoenle, E., et al. . . . 86
Manuel, A., s. Cohen, G., et al. . . . 203, 297
Marca, L., s. Fraccaro, M., et al. . . . 388
Marien, J., s. Fryns, J. P., et al. . . . 194
Martin, M. J., Abrisqueta, J. A.: Unusual pericentric inversion inv(9)(p13q11) in a phenotypically normal family (Cases observed) . . . 100
Martínez de Pancorbo, M., Isusquiza, M., Aquirre, A. I., Mazon, L. I., Lostao, C. M.: Red cell glyoxalase I polymorphism in Basque and Castilian populations (Orig. invest.) . . . 395
Mattei, J. F., s. Balestrazzi, P., et al. . . . 305
Mattei, J. F., Mattei, M. G., Giraud, F.: Prader-Willi syndrome and chromosome 15. A clinical discussion of 20 cases (Orig. invest.) . . . 356
Mattei, M. G., s. Balestrazzi, P., et al. . . . 305
Mattei, M. G., s. Mattei, J. F., et al. . . . 356
Matthews, K. B., s. Winter, R. M., et al. . . . 156
Mauff, G., Steuer, M., Weck, M., Bender, K.: The C4 β-chain: Evidence for a genetically determined polymorphism (Short comm.) . . . 186
Mayakowa, S. A., s. Puchkowa, G. P., et al. . . . 257
Mayer, H., Breyel, E., Bostock, C., Schmidtke, J.: Assignment of the human parathyroid hormone gene to chromosome 11 (Short comm.) . . . 283
Mazon, L. I., s. Martínez de Pancorbo, M., et al. . . . 395
McBride, W., s. Cohen, G., et al. . . . 203
McKay, E., s. Wieacker, P., et al. . . . 139
Metaxas, M., s. Schoenle, E., et al. . . . 86
Miguel, A., Ramon, M., Petitpierre, E., Goos, C. M. A. A., Vermeesch-Markslag, A. M. G., Vermorken, A. J. M.: Population screening for glucose-6-phosphate dehydrogenase deficiency on the Baleares (Orig. invest.) . . . 176
Mikkelsen, M., s. Brøndum Nielsen, K., et al. . . . 240
Minoda, K., s. Motegi, T., et al. . . . 160, 205
Mitchell, G., s. Frézal, J., et al. . . . 311
Morley, A. A., s. Dempsey, J. L., et al. . . . 288
Morton, N. E., Simpson, S. P.: Kinship mapping of multilocus systems (Orig. invest.) . . . 103
Motegi, T., Kaga, M., Yanagawa, Y., Kadowaki, H., Watanabe, K., Inoue, A., Komatsu, M., Minoda, K.: A recognizable pattern of the midface of retinoblastoma patients with interstitial deletion of 13q (Orig. invest.) . . . 160
Motegi, T., Komatsu, M., Minoda, K.: Is the interstitial deletion of 13q in retinoblastoma patients not transmissible? (Letter to the editor) . . . 205
Mowrey, P., s. Juberg, R. C., et al. . . . 216
Müller, C. R., Davies, K. E., Cremer, C., Rappold, G., Gray, J. W., Ropers, H. H.: Cloning of genomic sequences from the human Y chromosome after purification by dual beam flow sorting (Orig. invest.) . . . 110
Müller, Hj., s. Schmid, M., et al. . . . 363
Munnich, A., s. Frézal, J., et al. . . . 311

Nakasono, I., Iwasaki, M., Ogata, M., Yoshida, C., Yoshitake, T., Narita, K., Suyama, H., Tomohiro, Y.: A new single band variant of the Gc subtypes determined by isoelectric focusing (Short comm.) . . . 184
Narita, K., s. Nakasono, I., et al. . . . 184
Niebuhr, E., s. Tabor, A., et al. . . . 196
Niikawa, N., s. Iselius, L., et al. . . . 343
Nissimov, R., s. Shabtai, F., et al. . . . 273
Noel, B., s. Iselius, L., et al. . . . 343
Nomura, K., s. Yoshimitsu, K., et al. . . . 200
Nordmann, Y., s. Grandchamp, B., et al. . . . 180
Nussbaum, R. L., Airhart, S. D., Ledbetter, D. H.: Expression of the fragile (X) chromosome in an interspecific somatic cell hybrid (Orig. invest.) . . . 148

O'Brien, T. J., s. Wulfsberg, E. A., et al. . . . 271
Oehme, R., s. Horst, J., et al. . . . 263
Oehme, R., Kohne, E., Kleihauer, E., Horst, J.: Hb M Milwaukee: Direct detection of the β-globin gene mutation in three generations of an afflicted family (Orig. invest.) . . . 376
Ogata, M., s. Nakasono, I., et al. . . . 184
Ohama, K., s. Yoshimitsu, K., et al. . . . 200
Olaisen, B., s. Schamaun, O., et al. . . . 380

Page, L., s. Tantravahi, U., et al. . . . 33
Palmiter, R. D., s. Cox, D. R. . . . 61
Parker, J. W., s. Quattromani, F., et al. . . . 116
Pascali, V. L., Auconi, P.: Transferrin: Common and rare variants in Italy. Evidence for the existence of the rare $Tf^C 6$ among Caucasians (Orig. invest.) . . . 232
Pasquali, F., s. Iselius, L., et al. . . . 343
Patutschnick, W., s. Görg, A., et al. . . . 222

Pearson, P., s. Wieacker, P., et al. . . . 139
Pelz, L., s. Gottschall, A., et al. . . . 304
Pentz, S., s. Vogel, W., et al. . . . 246
Petersen, R., s. Dryja, T. P., et al. . . . 151
Peterson, I. S., s. Puchkova, G. P., et al. . . . 257
Petit, P., Fryns, J. P.: Two pericentric inversions inv(7)(p15;q32) and inv(9)(p11;q13) in a male with absence of vas deferens (Cases observed) . . . 303
Petitpierre, E., s. Miguel, A., et al. . . . 176
Phengsavath, H., s. David, V., et al. . . . 189
Poll, E. H. A., s. Kwee, M. L., et al. . . . 384
Postel, W., s. Görg, A., et al. . . . 222
Poulsen, H., s. Brøndum Nielsen, K., et al. . . . 240
Prigogina, E. L., s. Fleischman, E. W., et al. . . . 254
Prigogina, E. L., s. Puchkova, G. P., et al. . . . 257
Probeck, H. D., s. Iselius, L., et al. . . . 343
Propping, P., Friedl, W.: Genetic control of adrenergic receptors on human platelets. A twin study (Orig. invest.) . . . 105
Pteruschka, L., Machill, G., Wehnert, M., Seidlitz, G., Knapp, A.: Reliability of the Tønnesen technique for the identification of Hunter carriers (Short comm.) . . . 404
Puchkova, G. P., s. Fleischman, E. W., et al. . . . 254
Puchkova, G. P., Prigogina, E. L., Fleischman, E. W., Drosdova, T. S., Mayakova, S. A., Peterson, I. S.: Chromosome abnormalities in chronic myeloid leukemia in children (Orig. invest.) . . . 257

Quattromani, F., Shapiro, S. D., Young, R. S., Jorgenson, R. J., Parker, J. W., Blumhardt, R., Reece, R. R.: Clinical heterogeneity in the tricho-dento-osseous syndrome (Orig. invest.) . . . 116

Ramon, M., s. Miguel, A., et al. . . . 176
Rapaport, J. M., s. Dryja, T. P., et al. . . . 151
Rappold, G., s. Müller, C. R., et al. . . . 110
Rauscher, K. H., Bauchinger, M.: Chromosome aberrations induced in patients treated with chemotherapeutic drugs and irradiation for acute lymphatic leukemia (Orig. invest.) 73
Reece, R. R., s. Quattromani, F., et al. . . . 116
Reghis, A., Troungos, C., Lostanlen, D., Krishnamoorthy, R., Kaplan, J. C.: Characterization of weak alleles at the DIA_1 locus (Mustapha 1, Mustapha 2, and Mustapha 3) in the Algerian population (Orig. invest.) . . . 173
Reuser, A. J. J., s. Barneveld, R. A., et al. . . . 227
Ritter, M., s. Schoenle, E., et al. . . . 86
Robinson, A., s. Iselius, L., et al. . . . 343
Roncarati, E., s. Iselius, L., et al. . . . 343
Ropers, H. H., s. Müller, C. R., et al. . . . 110
Ropers, H. H., s. Wieacker, P., et al. . . . 139, 143
Rüdiger, H. W., Dreyer, M., Kühnau, J., Bartelheimer, H.: Familial insulin-resistant diabetes secondary to an affinity defect of the insulin receptor (Clinical case report) . . . 407

Sachs, E., s. Iselius, L., et al. . . . 343
Salk, D., s. Brackertz, M., et al. . . . 334
Sardemann, H., s. Tabor, A., et al. . . . 196
Scappaticci, S., s. Iselius, L., et al. . . . 343
Schamaun, O., Olaisen, B., Gedde-Dahl, T., Jr., Teisberg, P.: Genetic studies of an apoA-I lipoprotein variant (Orig. invest.) . . . 380
Schenker, T., s. Schoenle, E., et al. . . . 86
Schimmer, O., s. Abel, G. . . . 131
Schinzel, A., s. Fraccaro, M., et al. . . . 388
Schinzel, A., s. Schoenle, E., et al. . . . 86
Schmid, M., Müller, Hj., Stasch, S., Engel, W.: Silver staining of nucleolus organizer regions during human spermatogenesis (Orig. invest.) . . . 363
Schmid, W., s. Schoenle, E., et al. . . . 86
Schmidt, A.: Unilateral split hand in one of monozygotic twins (Clinical case report) . . . 301
Schmidtke, J., s. Mayer, H., et al. . . . 283
Schoenle, E., Schmid, W., Schinzel, A., Mahler, M., Ritter, M., Schenker, T., Metaxas, M., Froesch, P., Froesch, E. R.: 46,XX/46,XY chimerism in a phenotypically normal man (Clinical case report) . . . 86
Schmutz, S. M., Simpson, N. E.: Suggested assignment of peptidase S (PEPS) to 4p11→4q12 by exclusion using gene dosage, accounting for variability in fibroblasts (Orig. invest.) . . . 134
Schwinger, E., s. Fonatsch, C. . . . 39
Schwinger, E., s. Iselius, L., et al. . . . 343
Seger, R. A., s. Hitzig, W. H. . . . 207
Seidlitz, G., s. Pteruschka, L., et al. . . . 404
Seshadri, R. S., s. Dempsey, J. L., et al. . . . 288
Seyer, J., s. Kang, E. S., et al. . . . 80
Shabtai, F., Klar, D., Nissimov, R., Vardimon, D., Hart, J., Halbrecht, I.: A new familial "fragile site" on chromosome 16 (q23–24). Cytogenetic and clinical considerations (Orig. invest.) . . . 273
Shapiro, S. D., s. Quattromani, F., et al. . . . 116
Sharma, T., s. Das, B. C. . . . 249
Simoni, G., s. Fraccaro, M., et al. . . . 388
Simoni, G., s. Iselius, L., et al. . . . 343
Simpson, N. E., s. Schmutz, S. M. . . . 134
Simpson, S. P., s. Morton, N. E. . . . 103
Smith, K. D., s. Burk, R. D., et al. . . . 339
Sparkes, M. C., s. Sparkes, R. S., et al. . . . 146
Sparkes, R. S., Sparkes, M. C., Crist, M., Anderson, C. E.: Glyoxalase I "Null" allele in a new family: Identification by abnormal segregation pattern and quantitative assay (Orig. invest.) . . . 146
Sparkes, R. S., s. Wulfsberg, E. A., et al. . . . 271
Sperling, K., s. Zakrzewski, S., et al. . . . 55
Stallard, R., s. Juberg, R. C., et al. . . . 216
Stamberg, J., s. Burk, R. D., et al. . . . 339
Stasch, S., s. Schmid, M., et al. . . . 363
Steinbach, P., Barbi, G., Baur, S., Vogel, W.: Expression of the fragile site Xq27 in fibroblasts. II. Evidence for negative and positive clones from heterozygous females and possible relationship between frequency and phenotype (Short comm.) . . . 279
Steinbach, P., Djalali, M.: Ineffectivity of accessory bisatellited marker chromosomes in inducing meiotic nondisjunction (Short comm.) . . . 402
Stephens, J. D., Filly, R. A., Callen, P. W., Golbus, M. S.: Prenatal diagnosis of osteogenesis imperfecta type II by real-time ultrasound (Clinical case report) . . . 191
Steuer, M., s. Mauff, G., et al. . . . 186
Stewart, J., s. Fitzgerald, P. H., et al. . . . 128
Stührenberg, M.: De novo inversion 1, in amniotic, fluid cell cultures (Cases observed) . . . 412
Suckling, R. D., s. Fitzgerald, P. H., et al. . . . 128
Suyama, H., s. Nakasono, I., et al. . . . 184
Sykes, J. A., s. Wulfsberg, E. A., et al. . . . 271

Tabor, A., Andersen, O., Lundsteen, C., Niebuhr, E., Sardemann, H.: Interstitial deletion in the "critical region" of the long arm of the X chromosome in a mentally retarded boy and his normal mother (Clinical case report) . . . 196
Tager, J. M., s. Barneveld, R. A., et al. . . . 227
Tantravahi, U., Kirschner, D. A., Beauregard, L., Page, L., Kunkel, L., Latt, S.: Cytologic and molecular analysis of 46,XXq− cells to identify a DNA segment that might serve as a probe for a putative human X chromosome inactivation center (Orig. invest.) . . . 33
Takeoka, Y., s. Yoshimitsu, K., et al. . . . 200
Tegelaers, F. P. W., s. Barneveld, R. A., et al. . . . 227
Teisberg, P., s. Schamaun, O., et al. . . . 380
Teufel, M., Enders, H., Dopfer, R.: Consanguinity in a Turkish family with thrombocytopenia with absent radii (TAR) syndrome (Clinical case report) . . . 94
Todd, T. A., s. Kang, E. S., et al. . . . 80
Tommerup, N., s. Brøndum Nielsen, K., et al. . . . 240
Tomohiro, Y., s. Nakasono, I., et al. . . . 184
Tønnesen, T., Güttler, F., Lykkelund, C.: Reliability of the use of fructose 1-phosphate to detect Hunter cells in fibroblast-cultures of obligate carriers of the Hunter syndrome (Orig. invest.) . . . 371
Trautmann, T., s. Vogel, W., et al. . . . 246

Troungos, C., s. Reghis, A., et al. 173
Tuddenham, E. G. D., s. Winter, R. M., et al. 156
Turleau, C., Créau-Goldberg, N., Cochet, C., Grouchy, J. de: Gene mapping of the Gibbon. Its position in primate Evolution (Orig. invest.) 65

Ueda, K., s. Yoshimitsu, K., et al. 200
Ullrich, E., s. Fraccaro, M., et al. 388
Usui, T., s. Yoshimitsu, K., et al. 200

Valido, C. L., s. Juberg, R. C., et al. 216
Vardimon, D., s. Shabtai, F., et al. 273
Veenema, H., s. Iselius, L., et al. 343
Verheyen, K., s. Elsen, A. F. Van, et al. 235
Vermeesch-Markslag, A. M. G., s. Miguel, A., et al. 176
Vermorken, A. J. M., s. Miguel, A., et al. 176
Verneuil, H. de, s. Grandchamp, B., et al. 180
Vigi, V., s. Fraccaro, M., et al. 388
Vigi, V., s. Iselius, L., et al. 343
Vogel, W., s. Steinbach, P., et al. 279
Vogel, W., Trautmann, T., Hörler, H., Pentz, S.: Cytogenetic and biochemical investigations on fibroblast cultures and clones with one and two active X chromosomes of a 69,XXY triploidy (Orig. invest.) 246
Volkova, M. A., s. Fleischman, E. W., et al. 254
Volpato, S., s. Iselius, L., et al. 343

Warburton, D., s. Hook, E. B. 24
Watanabe, K., s. Motegi, T., et al. 160
Wauters, J. G., s. Elsen, A. F. Van, et al. 235
Weck, M., s. Mauff, G., et al. 186
Wegner, R.-D., s. Iselius, L., et al. 343
Wehnert, M., s. Pteruschka, L., et al. 404
Weidinger, S., s. Görg, A., et al. 222
Weil, D., s. Grandchamp, B., et al. 180
Welch, J. P., s. Iselius, L., et al. 343
Weser, J., s. Görg, A., et al. 222
Westermeier, R., s. Görg, A., et al. 222
Westerveld, A., s. Barneveld, R. A., et al. 227
Wettke-Schäfer, R., Kantner, G.: X-linked dominant inherited diseases with lethality in hemizygous males (Review article) 1
Wieacker, P., Horn, N., Pearson, P., Wienker, T. F., McKay, E., Ropers, H. H.: Menkes kinky hair disease: A search for closely linked restriction fragment length polymorphism (Orig. invest.) 139
Wieacker, P., Wienker, T. F., Dallapiccola, B., Bender, K., Davies, K. E., Ropers, H. H.: Linkage relationships between retinoschisis, Xg, and a cloned DNA sequence from the distal short arm of the X chromosome (Orig. invest.) 143
Wiedersberg, H., s. Gottschall, A., et al. 304
Wienker, T. F., s. Wieacker, P., et al. 139, 143
Willems, P. J., s. Elsen, A. F. Van, et al. 235
Winsor, E. J. T., s. Iselius, L., et al. 343
Winter, R. M., Tuddenham, E. G. D., Goldman, E., Matthews, K. B.: A maximum likelihood estimate of the sex ratio of mutation rates in haemophilia A (Orig. invest.) 156
Witkowski, R., s. Fraccaro, M., et al. 388
Wulfsberg, E. A., Carrel, R. E., Klisak, I. J., O'Brien, T. J., Sykes, J. A., Sparkes, R. S.: Normal superoxide dismutase-1 (SOD-1) activity with deletion of chromosome band 21q21 supports localization of SOD-1 locus to 21q22 (Orig. invest.) 271
Wunder, E., Fleischer-Reischmann, B.: Response of lymphocytes from Fanconi's anemia patients and their heterozygous relatives to 8-methoxy-psoralene in a cloning survival test system (Orig. invest.) 167

Yanagawa, Y., s. Motegi, T., et al. 160
Yoshida, C., s. Nakasono, I., et al. 184
Yoshimitsu, K., Hatano, S., Kobayashi, Y., Takeoka, Y., Hayashidani, M., Ueda, K., Nomura, K., Ohama, K., Usui, T.: A case of 21q− syndrome with half normal SOD-1 activity (Clinical case report) 200
Yoshitake, T., s. Nakasono, I., et al. 184
Young, K. E., s. Burk, R. D., et al. 339
Young, R. S., s. Quattromani, F., et al. 116

Zakrzewski, S., Koch, M., Sperling, K.: Complementation studies between Fanconi's anemia cells with different DNA repair characteristics (Orig. invest.) 55
Zhang, S., s. Iselius, L., et al. 343
Zuffardi, O., s. Fraccaro, M., et al. 388
Zuffardi, O., s. Iselius, L., et al. 343

Announcements 101, 413

Erratum 310

Indexed in current contents